DIAGNOSTIC METHODS FOR POMPE DISEASE AND OTHER LYSOSOMAL STORAGE DISEASES

Abstract

Provided are methods of screening subjects for lysosomal storage diseases, preferably glycogen storage diseases, using a tetrasaccharide as a biomarker. In a more preferred embodiment, subjects are screened for Pompe disease (*i.e.*, glycogen storage disease type II). Also provided are neonatal screening assays. The present invention further provides methods of monitoring the clinical condition and efficacy of therapeutic treatment in affected subjects. Further provided are methods of measuring a tetrasaccharide biomarker by tandem mass spectrometry, preferably, as part of a neonatal screening assay for Pompe disease.